



OTULIN gene

OTU deubiquitinase with linear linkage specificity

Normal Function

The *OTULIN* gene provides instructions for making a protein that helps regulate inflammation, which is part of the body's early immune response to foreign invaders. Inflammation occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight the invaders and facilitate tissue repair. Inflammation can be turned on by a cellular process called ubiquitination, in which molecules called ubiquitin are attached to certain proteins. When foreign invaders are recognized, chains of ubiquitin molecules linked end-to-end, called linear ubiquitin chains, are attached to particular proteins. The addition of these chains stimulates signaling pathways that result in inflammation. Once the infection is under control, the body stops the inflammatory response to prevent damage to its own cells and tissues. The OTULIN protein helps control inflammation by removing linear ubiquitin chains.

In addition to inflammation, the OTULIN protein is thought to be involved in regulating development before birth and controlling cell death. Researchers are working to understand the protein's role in these processes.

Health Conditions Related to Genetic Changes

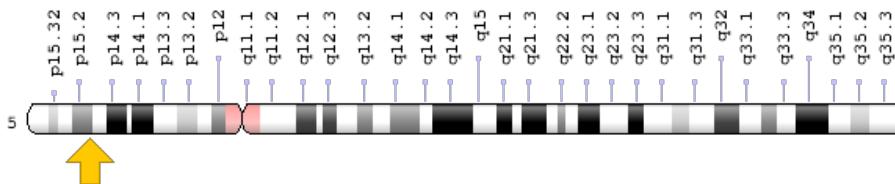
otulipenia

At least three mutations in the *OTULIN* gene have been found to cause otulipenia, a condition that causes abnormal inflammation in the body, beginning in infancy. Affected babies have recurrent episodes of fever, diarrhea, painful joints, and skin rashes. The gene mutations involved in otulipenia reduce the function of the OTULIN protein. As a result, removal of linear ubiquitin chains is impaired, and signaling pathways that cause inflammation are abnormally active. The excessive inflammation that results causes the signs and symptoms of otulipenia and damages organs in the body; it can be life-threatening if not treated.

Chromosomal Location

Cytogenetic Location: 5p15.2, which is the short (p) arm of chromosome 5 at position 15.2

Molecular Location: base pairs 14,660,794 to 14,716,552 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AIPDS
- deubiquitinating enzyme otulin
- FAM105B
- family with sequence similarity 105, member B
- FLJ34884
- GUM
- OTU domain-containing deubiquitinase with linear linkage specificity
- ubiquitin thioesterase Gumby
- ubiquitin thioesterase otulin

Additional Information & Resources

Educational Resources

- Informed Health Online (2006): What Happens When You Have an Inflammation?
https://www.ncbi.nlm.nih.gov/books/NBK279298/#_i2137_whathappenswhenyouha_

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28OTULIN%5BTIAB%5D%29+OR+%28OTU+deubiquitinase+with+linear+linkage+specificity%5BTIA%5D%29%29+OR+%28OTU+domain-containing+deubiquitinase+with+linear+linkage+specificity%5BTIAB%5D%29+OR+%28deubiquitinating+enzyme+otulin%5BTIAB%5D%29+OR+%28ubiquitin+thioesterase+Gumby%5BTIAB%5D%29+OR+%28ubiquitin+thioesterase+otulin%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- OTU DEUBIQUITINASE WITH LINEAR LINKAGE SPECIFICITY
<http://omim.org/entry/615712>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_OTULIN.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=OTULIN%5Bgene%5D>
- HGNC Gene Family: OTU domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/669>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25118
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/90268>
- UniProt
<http://www.uniprot.org/uniprot/Q96BN8>

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